IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)
e-ISSN: 2279-0853, p-ISSN: 2279-0861.Volume 20, Issue 9 Ser.10 (September. 2021), PP 43-46
www.iosrjournals.org

Two Interesting Cases of Blindness In Early Infancy

Dr. B Sreelatha[1], Dr. Pratibha[2]
[1](Senior Resident, Department of Pediatrics, Vinayaka Mission Medical College and Hospital, Karaikal)
[2](Junior Resident, Department of Pediatrics, Vinayaka Mission Medical College and Hospital, Karaikal)

Abstract:
The window of opportunity in treating a visually impaired infant is narrow and hence we pediatricians, general practitioners and midwives should be educated and encouraged to perform the red reflex test using direct ophthalmoscope. All health care personnel working for the care of the infant should be sensitized to the eye conditions in infancy and on the causes of childhood blindness and visual impairment. Here we present two case reports of blindness detected in the early infancy[1].
Case 1 is a 3-month-old infant upon a regular follow up was noticed to have unattained visual fixation. Ophthalmic evaluation revealed poor perception of light and projection of rays. Baby had poor response in bilateral occipital regions on doing a visual evoked response to brief flashes suggesting cortical visual impairment (CVI). MRI was normal. Management: visual stimulation therapy improved the eye fixation and child is currently on low visual aids.
Second case is a 1.5-month-old male child born to second degree consanguineous parents upon routine evaluation was found to be floppy with unattained visual fixation. Ophthalmic evaluation revealed bilateral macular cherry red spot narrowing down the diagnosis to inborn metabolic disorders (IMDS). Focused exome sequencing was done which showed polymorphism in Chr.15q23 suggestive of GM2 gangliosidosis. Hexosaminidase A enzyme assay showed deficiency of α subunit of the enzyme confirming the diagnosis. Management: Parents were explained about the overall prognosis and genetic counselling was offered.
Conclusion*: An early and timely detection of blindness in infancy period helps to intervene early which indeed is important in prevention and improving the visual outcome.*
Keywords*: Cortical visual impairment, Inborn metabolic disorders, GM2 gangliosidosis*

Date of Submission: 08-09-2021 Date of Acceptance: 23-09-2021

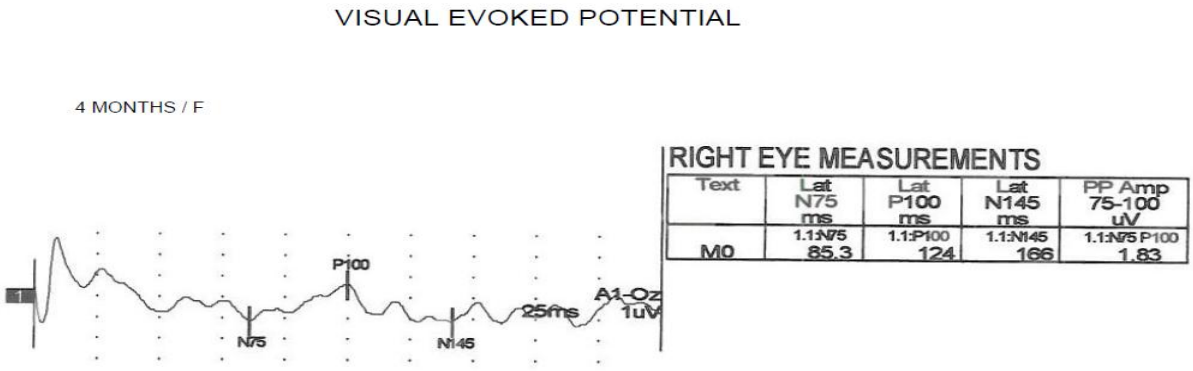
I. Introduction:

Blindness or severe visual impairment in infants and young children has an adverse outcome not only for vision but also on the growth, development, social and economic opportunities. Severe visual impairment and blindness should be recognized at the earliest and intervened whenever possible to retain the maximum possible vision. The causes of blindness in this age group maybe prenatal, perinatal or post-natal[1]. Congenital anomalies like enophthalmos, microphthalmos, coloboma, congenital cataract and infantile glaucoma must be identified by regular evaluation of eyeball at the time of birth and during subsequent visits so that maximum possible visual recovery can be restored. Ophthalmia neonatorum and retinopathy of prematurity have been identified as serious etiologies progressing to blindness for which special ophthalmic programs are currently available for screening, early detection and early access to therapy[2].Rare causes of blindness such as storage disorders or cortical visual impairment (earlier known as cortical blindness) are not identified adequately most of the times in the community until the baby presents with complete blindness[1].Here, we present two rare cases of blindness in early infancy. Though visual recovery is unlikely, early identification and treatment renders a possibility of delaying blindness and appropriate genetic counselling regarding future pregnancies.

II. Case Reports

CASE 1: A term female baby was delivered outside and brought at 2nd hour of life for complaints of weak cry. On admission, baby was asphyxiated with stable vital signs and subtle seizures. The nails and umbilical cord showed staining with meconium and the baby was SGA (asymmetrical IUGR) with birth weight of 2.01kg. Baby was admitted in NICU, given nutritional support and antibiotics and phenobarbitone for subtle seizures. Seizures were controlled with single anti-epileptic drug. Breastfeeding was initiated at 48 hours of life and then discharged from NICU on day 4 of admission. At discharge, on day 10, baby was active, tolerating exclusive breastfeeding, on tapering doze of phenobarbitone and plateauing of weight. USG cranium and OAE were normal and was hence, discharged. There was no dysmorphology noted. During follow up, child had normal development with normal head circumference. Though social smile developed, visual fixation remained

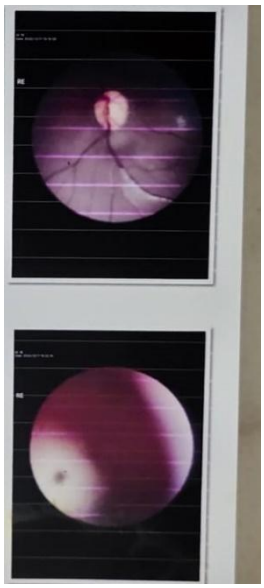
unattained at the end of three months. Ophthalmic evaluation was sought which revealed a normal cornea, conjunctiva, anterior chamber, vitreous, retina and optic nerve. However, perception of light and projection of rays (PL and PR) was recorded as poor, but not absent. The baby underwent visual evoked responses recording to brief light flashes. There were poor responses in bilateral occipital regions and possibility of cortical visual impairment (CVI) was suggested. MRI of brain was normal without structural defects of occipital lobes or features of hypoxic ischemic encephalopathy. During subsequent follow up, while on visual stimulation therapy, eye fixation developed at the end of 8 months. However, baby's visual acuity at one year was 3/60 in right eye and 2/60 in left eye. The child is presently 3 years and on low visual aids.

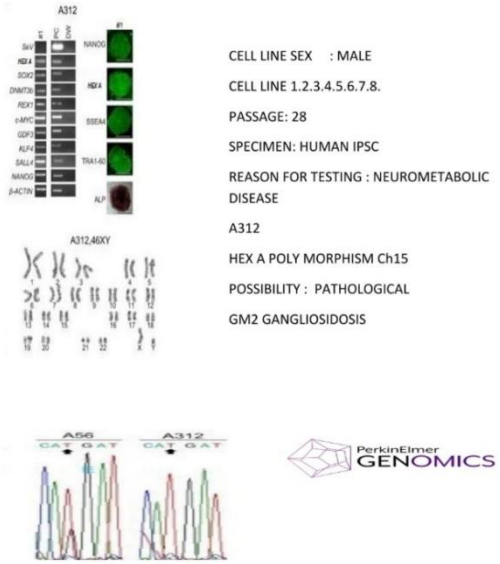


CASE 2: A 1.5-month-old male baby was born at term, third born to second degree consanguineous parents was brought for well-baby vaccination visit. There was a history of male eldest sibling death at 2.5 years of age. The cause was not evaluated. The second born female sibling is 2 years old, alive and healthy. During routine evaluation of the baby, it had not developed visual fixation. Baby was floppy with extreme head lag in pull-to-sit test and always presumed a frog leg posture. Head circumference was appropriate. There was no hepatosplenomegaly. The baby was at term outside through normal vaginal delivery without any history suggestive of asphyxia. There was no history of trauma to eye or any prolonged medications given to the baby. The baby was on exclusive breastfeeding and thriving well. Ophthalmic evaluation revealed normal cornea, anterior chamber, iris, lens bilateral pupils were equal and normally reactive to light. Fundus examination revealed bilateral macular cherry red spot. The differential diagnosis for cherry red spot is:

1. Gangliosidosis – GM1, GM2
2. Niemann Pick disease – A and B
3. Farber lipogranulomatosis
4. Metachromatic leukodystrophy
5. Galactosialidosis

Focused exome sequencing was done which revealed polymorphism in Chr.15q23 suggestive of GM 2 Gangliosidosis. Assay of α and β subunits of hexosaminidase A was done which revealed deficiency of α subunit of the enzyme. The visual and overall prognosis was explained to the parents and genetic counselling was offered.





Right eye Left eye Focused Exome Sequencing (FES) – Neuro metabolic sequencing

III. Discussion

Cortical visual impairment is increasingly becoming common in developing countries[3]. It is also referred to as cerebral visual impairment or neurologic visual impairment and is no more referred as cortical blindness. It is characterized by impairment in usual activities or visual field on the tasks guided by vision in which there is disorder in the post geniculate visual pathways of the brain and has no ocular pathology[4]. The most common on CVI is hypoxic ischemic damage in premature infants. Other causes include neonatal hypoglycemia, traumatic brain injury, hydrocephalus, structural brain anomalies and seizures. Screening and evaluation in young infants remain a challenge because of normal methods of assessing visual acuity and visual fields cannot be employed. In such situations, children need to be observed for their visual behavior over longer period of times in a dedicated environment. Recent use of preferential looking tests (PLT), especially forced PLT, has been used with some success[5]. Though visual evoked potential efficiency is a feature of higher cortical visual processing, it has high variability in children and is of limited use in the diagnosis of CVI. SWEEP Visual Evoked Potential (VEP) shows some promise as compared to FLASH VEP[6]. Structural defects in brain usually accompany CVI. However, a normal MRI of the brain does not rule out CVI. In such cases, functional MRI (fMRI) or PET Scan of brain, diffused tensor MRI may be helpful[7]. The best management for cortical visual impairment is prevention of premature births or at least effective management of babies born pre-term, protecting them from hypoxic brain injuries. Stem cell therapy and various modes of visual stimulation are still in infant stage of routine management protocol[8].

Inborn metabolic disorders (IMDS) are a rare group of disorders of metabolic pathway with heterogeneous presentations affecting various systems of the baby. IMDS has an underlying genetic defect associated. Of all the systems involved, ocular involvement plays a very crucial role as they guide to the diagnosis of specific IMD. Narrowing down the spectrum of IMD after initial ophthalmic evaluation, it becomes easy to get genetic analysis which otherwise would cost the patient heavily. Early diagnosis is mandatory because IMDs are rapidly progressive and cause irreversible damage. Whenever treatment is available, the long-term outcome will be good only if commenced early. Identification of particular IMD helps in genetic counselling[9]. Though newborn screening is an ideal way to diagnose early in resource poor countries, at least a late identification can be avoided when early markers are picked in well-baby / high risk baby visits. Even today, recording a good clinical history, thorough physical examination, early identification of developmental deviation, appropriate investigation and accurate diagnosis serve as a gold standards in office practice[10].

Referrences

[1]. Gogate P, Gilbert C, Zin A. Severe visual impairment and blindness in infants: Causes and opportunities for control. Middle East Afr J Ophthalmol 2011; 18:109-14.
[2]. Gogate P, Deshpande M, Sudrik S, Kishore H, Taras S, Deshpande M. changing pattern of childhood blindess in Maharashtra, India. Br J Ophthalmol 2007; 91:8-12.
[3]. Bodeau-Livinec F, Surman G, Kaminski M, et al. Recent trends in visual impairment and blindness in the UK. Archives of Disease in Childhood 2007; 92:1099-1104.
[4]. Good WV, Jan JE, DeSa L, et al. Cortical Visual Impairment in Children. SurvOphthalmol. 1994; 38(4):351-64
[5]. Dr Satish Thomas. Cortical visual impairment. IAP e-newsletter - Child India, August 2021:38-39

[6]. Good WV. Development of a quantitative method to measure vision in children with chronic cortical visual impairment. Trans Am Ophthalmol Soc. 2001; 99:253-69
[7]. Orbitus E, Verhoeven J, Sunaert S, et al. Integrity of inferior longitudinal fasciculus and impaired object recognition in children: a diffusion tensor imaging study. Dev Med Child Neurol. 2012. 54(1):38-43
[8]. Chang MY, Borchert MS. Advances in the evaluation and management of cortical/cerebral visual impairment in children. SurvOphthalmol 2020. 65:708-724
[9]. Poll-The BT, Maillette de Buy Wenniger-Prick LJ, Barth PG, Duran M. The eye as a window to inborn errors of metabolism. J Inherit Metab Dis. 2003;26(2–3):229–44.
[10]. Neha Gandhi Madan. Typical ophthalmic features- road to diagnose inherited metabolic disorder. IAP e-newsletter - Child India, August 2021:28-31.

Dr. B Sreelatha, et. al. "Two Interesting Cases of Blindness In Early Infancy."*IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)*, 20(09), 2021, pp. 43-46.